

## **PARENTS GUIDE TO DELETION 22Q11**



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This booklet aims to talk you through some of the areas which **may** affect your child. However, if you have any specific queries then please do not hesitate to contact the foundation.

<b>TOPIC</b>	<b>PAGE NUMBER</b>
WHAT IS VELO-CARDIO-FACIAL SYNDROME?	3-5
EVALUATIONS AT INITIAL DIAGNOSIS TO ESTABLISH THE EXTENT OF ANOMALIES	5-6
WHAT DOES HAVING A DELETION MEAN?	6-7
HOW IS THIS DELETION DETECTED?	7
WHY ARE THERE SO MANY NAMES?	7
SYNDROME VS SEQUENCE	7
LONG TERM PROGNOSIS	8
WHAT ARE THE CHANCES OF THIS RECURRING?	8
MEDICAL SECTION	9
• CAROTIDS ARTERIES	9
• CALCIUM	9
• CONSTIPATION	9
• DENTAL	9
• EYES (VISION)	10
• FACIAL FEATURES	10
• HEART	11
• HERNIAS	11
• HEARING/EARS	11-12
• HYPOSPADIAS	12
• IMMUNE SYSTEM	12
• KIDNEY DEFECTS	12-13
• LEG PAINS	13
• PALATE	13-14
• SPINA BIFIDA OCCULTA	14
DEVELOPMENTAL DELAY	14-15
SPEECH AND LANGUAGE	16
EDUCATION AND SCHOOLING SECTION	16-17
• EARLY INTERVENTION	17
• PRESCHOOL	17
• SCHOOL	17-18
• INDIVIDUAL EDUCATION PLAN (IEP)	18
SERVICES TO HELP WITH LEARNING NEEDS	18
• TUTORING	18
• LEARNING LINKS	18
• CHERI	19
LEAVING SCHOOL	19
EMPLOYMENT	19
AS ADULTS	20
MENTAL HEALTH ISSUES	20
TELLING YOUR CHILD THEY HAVE VCFS	21
BENEFITS	21-22
HOW DO I CLAIM THE ABOVE PAYMENTS?	22

## WHAT IS VELO-CARDIO-FACIAL SYNDROME?

Velo-cardio-facial syndrome is a genetic syndrome. It is the result of a submicroscopic deletion on the long arm of Chromosome 22 in the “q11” region. Most cases of VCFS arise due to the deletion occurring sporadically i.e. for no apparent reason. This means no previous family member was affected. Other cases occur due to the deletion being passed on by a parent with it, this is called autosomal dominant inheritance.

VCFS is much more common than previously thought. It is currently estimated that around 1 in 2 - 4,000 births are affected making it the second most common genetic syndrome (Gothelf & Lombroso, 2001). Due to the variable expression of VCFS the incidence is probably much higher than previously estimated and thus this rate is constantly being reviewed. It is the most common cause of congenital heart defects after Down syndrome.

The name Velo Cardio Facial Syndrome describes the three most common features of the syndrome.



Velo – (or velum) means the palate.  
Cardio – means that the heart is affected.  
Facial – VCFS people have similar facial features.  
Syndrome – is a collection of similar findings.

VCFS is more than just these three features. Below are the characteristic findings.

### **Congenital heart disease**

- Found in about 74% of affected individuals
- Usually conotruncal malformations
  - Teratology of Fallot
  - Interrupted aortic arch
  - Truncus arteriosus
  - Ventricular septal defect (VSD)
- Conotruncal malformations identified in infancy/early childhood and can be surgically repaired
- Mild problems with the aorta or surrounding venous system may not be recognized

### **Palatal abnormalities**

- Found in about 69% of affected individuals

- Several types of abnormalities observed
  - Velopharyngeal incompetence (VPI)
  - Submucosal cleft palate
  - Visible cleft palate
  - Cleft lip and palate (rare)
  - Bifid uvulae

- May be surgically repaired
- May affect speech development
- Speech therapy is recommended

### **Feeding difficulties**

- Found in about 30% of affected individuals
- May be due to palate or heart problems
- May also be caused by problems transporting food from the mouth to the esophagus
- Signs of feeding difficulty
  - Infants are irritable, gag, vomit, have a weak suck
  - Younger children find spoon feeding easier but have trouble drinking from a cup
  - Older children have difficulty with lumpy, crunchy foods
  - May be treated by teaching children better feeding practices

### **Immunodeficiency**

- Caused by small or missing thymus gland
- Causes greatest number of problems in first year of life
- Children may have frequent infections such as otitis media, bronchitis or pneumonia

### **Hypoparathyroidism**

- Regulates calcium levels in bloodstream
  - Absence or underdevelopment leads to hypocalcemia
  - May cause tremors, seizures, muscle spasms, abnormal breathing, and abnormal heart rhythms
- Diagnosed by a blood test
- Treated with calcium supplements

### **Growth hormone deficiency**

- Approximately 41% of affected individuals are below the 5<sup>th</sup> percentile in height
- May be caused by abnormalities in formation of pituitary gland
  - In only about 1/10 of individuals with 22q11 deletion
  - Causes reduced levels of growth factors
- Can be determined with a blood test
- Treatment with injections of growth hormones

### **Characteristic Facial Features**

- Nose - bulbous nasal tip, prominent nasal root, nasal dimple
- Ear - overfolded helices; cupped, microtic, and protuberant ears; narrow external auditory meati
- Eyes - hooding of upper or lower lid, ptosis, epicanthal folds

### **Renal anomalies**

- Found in about 37% of affected individuals
- Includes single kidney, echogenic kidney, small kidneys, and other renal anomalies

### **Autoimmune disorders**

- Juvenile rheumatoid arthritis (JRA)
  - About 20 times more frequent than in the general population
  - Age of onset is 17 months to 5 years
- idiopathic thrombocytopenia purpura (ITP)
  - seen 200 times more frequently than in the general population
- hyperthyroidism (Grave's disease),
- hypothyroidism
- vitiligo (skin condition that causes loss of pigment, resulting in irregular pale patches of

skin).

- hemolytic anemia
- autoimmune neutropenia
- aplastic anemia
- celiac disease

### **Developmental difficulties**

- Found in over 90% of affected individuals
- Delayed development of motor skills
  - Due to problems with muscle strength and coordination
  - Often delayed in sitting, crawling, walking
  - May be helped by physical therapy
- Learning
  - Typically have slower learning rate
  - Follow same learning sequence as other children but at slower pace
  - Often have strong verbal skills but weaker math skills
  - May have difficulty paying attention
- Early diagnosis and intervention important
- May require learning support

### **Speech and language**

- Verbal speech usually does not develop until 2-3 years
- May also have difficulty in formation of sounds
- Palatal problems often lead to speech problems
- Early intervention by speech and language pathologist is important

### **Behavioral difficulties**

- About 30% of affected individuals develop psychiatric illness
  - Depression
  - Anxiety
  - Schizophrenia
  - Bipolar disorders
- May be evident as tantrums and mood swings in childhood
- Social skills training and counseling may be necessary

**See our website for the downloadable pdf Specialist Fact Sheet listing the 186 reported anomalies.**

## **EVALUATIONS AT INITIAL DIAGNOSIS TO ESTABLISH THE EXTENT OF ANOMALIES**

In the neonatal period:

- Measurement of serum calcium concentration to assess for hypoparathyroidism, followed by a formal endocrinology evaluation if abnormal
- Measurement of absolute lymphocyte count; a low absolute lymphocyte count necessitates evaluation of T- and B-cell subsets and referral to an immunologist.
- Renal ultrasound examination to evaluate for structural renal defects
- Chest x-ray to evaluate for thoracic vertebral anomalies
- A baseline cardiac evaluation by a cardiologist that includes a chest x-ray, ECG and echocardiogram; if a vascular ring is suspected, a chest MRI may be required.
- Have the palate examined for clefting or submucous clefting.
- Have an eye examination

In infancy:

- Assessment for possible feeding problems such as significant gastroesophageal reflux; difficulty with sucking/swallowing, advancing feeds, addition of textured foods; vomiting and constipation
- Speech and language assessment by age two given that almost all affected children have delay in emergence of language and would benefit from early intervention strategies. In addition, such an evaluation aids in the diagnosis of a palatal abnormality/VPI.

After age four:

- Yearly review by your paediatrician / general practitioner  
As needed:
- Evaluation of children with short stature (height below the 2nd centile) by an endocrinologist for possible growth hormone deficiency
- Evaluation in any person with evidence of anxiety, mood disorder, behavioral differences, or frank psychoses
- Evaluation by a hematologist of any person with a history of a bleeding disorder
- Evaluation by an immunologist of any person with serious recurrent infections

### **Ongoing Surveillance**

Affected individuals require follow-up as needed on a "system by system" basis. For example if a cleft palate has been repaired follow-up by the cleft palate team is required, heart defects both corrected and uncorrected also need to be monitored by a cardiologist.

Regular developmental assessments benefit the child and assist the school in providing appropriate remediation.

Periodic re-evaluation by a medical geneticist can apprise the family of new developments and/or recommendations.

## **WHAT DOES HAVING A DELETION MEAN?**

Human cells contain the genetic information which is required to direct development and maintenance of the body. This information is carried within the genes. Genes may be viewed as instructions to the cell as to how to behave. The genes are carried on chromosomes, each cell having 23 pairs of chromosomes, one of each pair originating from each parent and transmitted in the sperm or egg. One pair will be the sex chromosomes, the X and the Y chromosomes; individuals with an X and a Y chromosome are male, individuals with two X chromosomes are female. The remainder of the chromosomes are named by number 1 through 22.

Occasionally a mistake is made when the chromosomes are packaged into the sperm or egg and a child is conceived. Most of us know of Down syndrome (also known as Trisomy 21). This is the result of the wrong number of chromosomes being packaged together and shows the importance that the current number of chromosomes is needed for normal development.

In VCFS instead of having an extra chromosome, the correct number are present however part of chromosome 22 is missing (deleted). Under a microscope Chromosomes are seen to have two arms and a striped appearance. The long arm is referred to as 'q' and the short arm is referred to as 'p'. The dark and light stripes are called bands. A missing band, or part of a band is termed a deletion. The deletion in VCFS involves band 11 on the long arm of chromosome 22. Therefore this chromosome abnormality is sometimes

referred to as the 22q11 deletion syndrome. 22q11 referring to the chromosome and area of the missing genes. From this, we can conclude that VCFS is a disorder caused by abnormal gene dosage (in this case too small a dose of genes).

Genetic conditions cannot be cured because faults in the genes cannot be fixed. Genetic research continues at a rapid rate but it is not appropriate to expect a cure the condition as a whole. Some defects can be corrected with surgery for example repairing a cleft palate or closing a heart defect such as a ventricular septal defect.

## **HOW IS THIS DELETION DETECTED?**

A test known as FISH (Fluorescence In Situ Hybridisation) is used. Scientists developed "DNA markers" which originate from 22q. These markers can be chemically manipulated so that they fluoresce under microscopic illumination, and are called probes. If added to chromosomes from an individual's cells they will stick to the part of chromosome 22q11 from which they originated. So, someone with VCFS and a deletion will only show one signal (on the undeleted chromosome 22), whereas an unaffected person will have two signals, one from each chromosome 22. FISH testing for Deletion 22q11 was introduced in 1994 and is now widely available for the clinical and prenatal diagnosis of deletion 22q11.

## **WHY ARE THERE SO MANY NAMES?**

There has been and continues to be much confusion over the name of this syndrome. Depending upon where and by whom your child was diagnosed, the syndrome is given a variety of names, e.g. Velo-Cardio-Facial Syndrome (VCFS), DiGeorge Syndrome (DGS), Shprintzen Syndrome, 22q11 Deletion, CATCH 22, conotruncal anomaly face syndrome and Cayler cardiofacial syndrome.

In clinical genetics syndromes are named in a number of different ways including:

- after those who first describe them
- symptomatically by the use of several key findings which appear frequently in affected individuals
- using acronyms
- using the first letters of clinical features associated with the syndrome.
- By the type chromosome rearrangement
- By the first known patient or the place or institution where it was first described.

There is no general consensus on the best naming system and as a result some syndromes are known by more than one name as is the case with VCFS. The origin of the names are briefly explained below.

For the purpose of this booklet the condition will be referred to as VCFS or Velo-Cardio-Facial Syndrome.

## **SYNDROME VS SEQUENCE**

There have been many reported sequences within the VCFS phenotype including:

- DiGeorge
- Robin (also referred to as Pierre Robin sequence)
- Potter

- CHARGE
- Holoprosencephaly

The difference between a syndrome and a sequence is a syndrome is a cluster of congenital abnormalities that occur because of a specific cause. A single causative factor, such as a genetic deletion, causes a series of anomalies, all of which can be traced back to this causative factor. A sequence is a collection of abnormalities that occur because of one leading to the next. In a sequence, multiple anomalies can be traced back to one anomaly which interfered with subsequent development.

(For a more in depth review of the above please refer to the an article written by Dr Robert Shprintzen, The Name Game. This can be found at [www.vcfsef.org](http://www.vcfsef.org)).

## **LONG TERM PROGNOSIS**

Providing your child does not have a severe, life threatening medical condition (e.g. heart, immune system) then their life expectancy is normal.

Given the availability of special education provision, young children should benefit substantially and will be able to reach their potential. Whether they will be able to live independently or not depends upon the individual ability to cope.

## **WHAT ARE THE CHANCES OF THIS RECURRING?**

Anyone with the deletion has a 50% chance of passing this deletion onto their children. That is a one in two chance for **each** pregnancy. It does not mean that if you have one affected child the next one will be unaffected as every pregnancy has the same 50% chance. As VCFS is a variable expression syndrome the child will not necessary be affected the same as the individual passing it on.

If you and your partner have been tested and do not have the deletion then the chances of you having another child with the deletion are low. It is said to be slightly higher than the general population as it has happened once but it is still low. There are two tests using FISH analysis available to check for VCFS during pregnancy. These are chorionic villus sampling (CVS) or amniocentesis.

Preimplantation genetic diagnosis (PGD) are available for families in which the diagnosis of Deletion 22q11 has been established in an affected family member. For laboratories offering PGD contact your local genetic service.

Genetic counsellors are available to speak about any risks of the deletion recurring or any other concerns you may have. Your local doctor can give you the name and number of a genetic counsellor close to you.

**REMEMBER: THERE IS NOTHING YOU DID TO CAUSE VCFS. IT OCCURS AT CONCEPTION AND NOTHING DONE DIFFERENTLY COULD HAVE CHANGED THE OUTCOME.**

# **MEDICAL SECTION**

Depending on the age and presenting problems of the child, a multidisciplinary evaluation involving healthcare providers from the following specialties is often necessary: genetics, plastic surgery, speech pathology, otolaryngology, audiology, dentistry, cardiology, immunology, child development, child psychology, neurology and general paediatrics. Some patients also require evaluation by healthcare providers specializing in feeding, endocrinology, rheumatology, gastroenterology, neurosurgery, general surgery, orthopaedics, urology, haematology, psychiatry, and ophthalmology.

This section contains some of the more frequently noted anomalies in alphabetical order.

## **CAROTIDS ARTERIES**

Carotid arteries are arteries that supply the brain with blood. In VCFS these arteries can be displaced. This can be a problem for children undergoing surgery in the throat region.

## **CALCIUM**

Many VCFS children are born with low blood calcium levels (hypocalcaemia). This is caused by hypoparathyroidism (underactive adrenal gland) which results in the hypocalcaemia. Calcium is essential to maintaining total body health. Your body needs it to keep your bones and teeth strong and to ensure proper functioning of muscles and nerves. Problems with the parathyroid gland normally correct themselves with age although recurrence of hypocalcaemia in later childhood has been reported. For this reason some doctors recommend annual calcium checks. Some children with hypocalcaemia have been known to suffer from hypocalcaemic fits so any kind of fit/seizure should be followed up with a calcium check.

## **CONSTIPATION**

A large number of children with VCFS are found to suffer from constipation. Constipation is passage of small amounts of hard, dry bowel movements, usually fewer than three times a week. People who are constipated may find it difficult and painful to have a bowel movement. Other symptoms of constipation include feeling bloated, uncomfortable, and sluggish. See your Doctor for treatment options.

## **DENTAL**

Some children with VCFS display certain dental characteristics such as higher number of cavities or poor quality teeth, which may decay quickly. These dental problems are fairly common in the primary (baby) dentition and much less common in the permanent (secondary) dentition.

If decay occurs because of congenital abnormalities of the teeth, don't worry too much. Have your dentist treat the teeth appropriately and remember that in most cases the secondary teeth will be fine.

## EYES (VISION)

People with VCFS often have vision or eye problems, many of which are readily improved with simple treatments, like exercises. The difficulties are often related to the low muscle tone commonly seen in VCFS, as the eyes rely on numerous sets of muscles to keep them functioning properly.

Typical vision findings amongst the VCFS population may include one or more of the following:

- Reduced accommodation (focussing).
- Reduced convergence (ability to point the eyes closely).
- Irregular retinal vessels.
- Squint.
- Puffy eyelids.
- Narrow eyelid openings.
- Poor visual perceptual function eg: memory encoding and reduced visual spatial skills.

It is important to include regular visual assessment as part of the ongoing care regimen for every person affected by VCFS.

## FACIAL FEATURES

Children with VCFS have very typical facial features which are often quite subtle in the individual and more pronounced in a group.

- As babies, the children tend to be very 'pretty' with small features including small ears (in relation to the rest of the family), small mouth and almond shaped eyes.
- The nose is often wider than the usual 'button nose' seen on most babies. When they get older their faces tend to become longer and the nose broadens at the tip and becomes 'bulbous'.
- VCFS children often display a lack of facial expression. This is due to low muscle tone of the facial muscles.
- Red, dark circles are sometimes seen under the eyes and are referred to as allergic shiners.
- Their eyelids often look puffy, particularly when they are babies and toddlers. This seems to disappear with age.



## HEART

It has been reported that approximately 74% of children with VCFS have some kind of congenital heart defect and this is often of a serious nature. A condition is called congenital when it is present at birth. Congenital Heart defect means the heart has not formed correctly. Some heart defects are of a serious nature and require surgical treatment at a very young age while others may never require surgery.

The heart defect may be diagnosed if the doctor hears an abnormal sound, referred to as a murmur. Children with normal hearts also can have heart murmurs. These are called "innocent" or "functional" murmurs. A doctor may suggest tests to rule out a heart defect. These tests may include an ECG (electrocardiogram), which assesses the heart's pacemaker (electrical) system and/or an echocardiogram. An echocardiogram is a specialized ultrasound of the heart that assesses the structure and function of the heart. If an echocardiogram shows that a heart defect is present then additional testing may be required. The cardiologist will discuss this with you if it is necessary.

Children and adults with certain heart defects, even after surgical repair, remain at increased risk of infection involving the heart and its valves (infective endocarditis). Parents of children with heart defects and adults with repaired heart defects should discuss with their doctor whether they need to take antibiotics before certain dental and surgical procedures in order to prevent these infections.



## HERNIAS

Hernias are common in the general population but are more so in children with VCFS. A hernia is a weakness or defect in the lining of the abdominal (belly) or pelvic (groin) wall. It can be present at birth or develop over the years. A hernia repair is usually done as day surgery and your child is up and about that evening.

## HEARING/EARS

Many children with VCFS suffer from frequent ear infections (otitis media). Otitis media means inflammation of the middle ear. The inflammation occurs as a result of a middle ear infection. It can occur in one or both ears. It is also the most common cause of hearing loss in children. It is serious because of the severe earache and hearing loss it can create. Hearing loss, especially in children, may impair learning capacity and even delay speech development. However, if it is treated promptly and effectively, hearing can almost always be restored to normal. Without proper treatment, damage from an ear infection can cause chronic or permanent hearing loss. If the doctor suspects your child has hearing loss they may refer you to an audiologist who may perform two tests, an audiogram and a

tympanogram for more information.

Otitis media may recur as a result of chronically infected adenoids and tonsils. If this becomes a problem, your doctor may recommend removal of one or both. Research has found that **“Removal of adenoids in children with VCFS is NOT recommended as this may worsen the VPI they already have by increasing the area in the back of the throat. This makes speech more difficult due to air escape through the nose”**. We therefore recommend you seek the services of a professional who is familiar with VCFS before having this surgery performed.

Many children with cleft palates have unusual anatomical differences in the ear, including narrow eustachian tubes which prevent the passing of fluids from the ears therefore the insertion of grommets is often performed even without evidence of otitis media as a preventative strategy.

## **HYPOSPADIAS**

This is a common defect that affects one in every 200 male babies. It is even more common in VCFS male infants. Hypospadias is a birth defect of the penis that commonly has four characteristics:

- The urethral opening is located on the underside of the penis, instead of the tip, and may exit the penis anywhere along its shaft as high as the scrotum.
- The urethral opening is unusually narrow.
- The entire foreskin may be bunched on the topside of the penis.
- The penis itself may be curved to one side.

Most often, hypospadias is noticed at birth; however, if the abnormalities are particularly mild, diagnosis may come later in life.

## **IMMUNE SYSTEM**

Some children with VCFS are born with an absent or partially absent thymus gland. The thymus is a ductless gland that plays an important role in the development of the immune system in early life, and its cells form a part of the body's normal immune system. If absent, some of the white cells (known as T cells) which are important in fighting infection, may not be as plentiful in the blood stream as they should be, and may not work as well. A blood test can help to find out whether this is the case. Children with low numbers of T cells can get more than their fair share of chest infections or diarrhoea. Occasionally they may get unusual or serious infections. The chances of this happening can be lowered by giving regular antibiotics, and in some cases by giving doses of antibodies (intravenous immunoglobulin) into a vein.

If your child has suspected immune system abnormalities we recommend that you consult an immunologist for advice about assessing the immune system, and vaccination.

## **KIDNEY DEFECTS**

A number of kidney defects are associated with VCFS. These include the kidneys may be in the wrong place (ectopia), in the wrong position (malrotation), joined together (horseshoe kidney), or missing (kidney agenesis). Polycystic kidney disease where the kidney contains many cysts (fluid-filled sacs) is also common.

While some kidney defects cause frequent urinary tract infections some do not cause symptoms and are never detected. Some untreated defects may interfere with the function of the kidneys, leading to kidney failure, which can require dialysis or kidney transplantation. Therefore all children with VCFS should have an ultrasound scan of the kidneys whether or not they are having any frequent bladder infections to ensure that there is no underlying problem.

## **LEG PAINS**

This was accepted as an anomaly due to research carried out by Ahmad Al-Khattat at University College, Northampton and instigated by The English 22q11 Group in 1995. It was reported that over 50% of children with VCFS have been found to suffer from leg pains. The pains were most commonly in the back of the leg, experienced daily and usually lasted less than one hour. The leg pains are associated with sleep disturbance and resulted in limited exercise tolerance. However, it was also found that these same children may also have no difficulty with interesting exercise activities.

The research also reported that foot abnormalities seem to be more common in VCFS patients than in the general population. Reported abnormalities include flat feet and tight Achilles tendons. Treatment for these conditions include cushioned shoe insoles which are designed according to the type of foot. This treatment improved leg pain, sleep disturbance and exercise difficulty in a significant number of VCFS children. It is therefore recommended that you consult a podiatrist if one or more of the above symptoms are experienced.

## **PALATE**

This is one of the most common areas where an anomaly is noted. Submucous clefts are the most commonly seen clefts in VCFS followed by overt cleft palates (hard/soft). Cleft lips have also been reported but are not as commonly seen. Velo Pharyngeal Insufficiency (VPI) is extremely common in VCFS.

### **WHAT IS A CLEFT PALATE?**

The word "palate" refers to the roof of the mouth and the term "cleft" indicates a split in the palate. The palate consists of both a bony portion (hard palate) and a muscular portion (soft palate). At the end of the soft palate, the small finger-like projection of tissue that hangs down is called the "uvula". A cleft palate is an opening in the roof of the mouth in which the two sides of the palate did not fuse, or join together, as the unborn baby was developing. This cleft can affect just the hard palate, just the soft palate or both (full cleft palate). A cleft lip is a separation of the two sides of the lip. The separation often includes the bones of the upper jaw and/or upper gum. Cleft lip and cleft palate can occur on one side (unilateral cleft lip and/or palate), or on both sides (bilateral cleft lip and/or palate). Because the lip and the palate develop separately, it is possible for the child to have a cleft lip, a cleft palate, or both cleft lip and cleft palate.

A child born with a cleft frequently requires several different types of services, e.g., surgery, dental/orthodontic care, and speech therapy, all of which need to be provided in a coordinated manner over a period of years. This coordinated care is provided by interdisciplinary cleft palate/craniofacial teams comprised of professionals from a variety of health care disciplines who work together on the child's total rehabilitation.

The good news is that both cleft lip and cleft palate are treatable birth defects. Most

children born with either or both of these conditions can have reconstructive surgery while they're still infants to correct the defect. An immediate problem after birth, however, is feeding, but special teats, bottles and even prostheses are available to ensure that children with oral clefting receive adequate nutrition until surgical treatment is provided.

## **WHAT IS A SUBMUCOUS CLEFT PALATE?**

A submucous cleft palate is one type of cleft palate. The term "submucous" refers to the fact that the cleft is covered over by the lining (mucous membrane) of the roof of the mouth. This covering of mucosa makes the cleft difficult to see when looking in the mouth.

A submucous cleft of the soft palate is characterized by a midline deficiency or lack of muscular tissue and incorrect positioning of the muscles. A submucous cleft of the hard palate is defined as a bony defect in the midline or center of the bony palate. This can sometimes be felt as a notch or depression in the bony palate when the palate is palpated with a finger. Often a submucous cleft palate is associated with a bifid or cleft uvula.

Anyone diagnosed with VCFS should be assessed by a cleft palate team. Your Doctor can write a referral for you to attend your nearest clinic.

## **SPINA BIFIDA OCCULTA**

Spina bifida is a type of neural tube defect. Spina bifida (which is Latin for "split spine") is a developmental birth defect involving the neural tube: incomplete closure of the embryonic neural tube results in malformed vertebrae that do not fully enclose the spinal cord. Spina bifida malformations fall into three categories: spina bifida occulta, spina bifida cystica (myelomeningocele), and meningocele. It is spina bifida occulta that is associated with VCFS. Occulta is Latin for "hidden." This is the "mildest" form of spina bifida although the degree of disability can vary depending upon the location of the lesion and actually be very severe in some patients.

In occulta there is no opening of the back, but the outer part of some of the vertebrae are not completely closed. The split in the vertebrae is so small that the spinal cord does not protrude. The skin at the site of the lesion may be normal, or it may have some hair growing from it; there may be a dimple in the skin, or a birthmark. People with this form may have no symptoms or problems while others may have incontinence, ambulatory problems, loss of sensation, deformities of the hips, knees or feet and loss of muscle tone. Depending on the location of the lesion, intense pain may occur originating in the lower back, and continuing down the leg to the back of the knee. Many people with the mildest form of this type of spina bifida do not even know they have it, or symptoms do not appear until later in life.

## **DEVELOPMENTAL DELAY**

Some form of developmental delay is present in over 95% of individuals with VCFS. Not all VCFS individuals will be delayed to the same degree. Some fall into the borderline/mild category while others the severe category. A developmental delay is present when your child has the delayed achievement of one or more of their milestones. Milestones are behaviors that emerge over time, forming the building blocks for growth and continued learning. Some of the categories within which these behaviors are seen include:

- **Cognition** (thinking, reasoning, problem-solving, understanding)

- **Language/speech** (expressive and receptive abilities)
- **Motor coordination** (gross/fine motor, jumping, hopping, throwing/catching, drawing, stacking)
- **Social interaction** (initiating peer contact, group play)
- **Self-help** (dressing, eating, washing).

Your Pediatrician should screen for delays at your child's checkups. This may consist of simple questions to see what your child is able to do at different ages, or it may include a formal developmental assessment. These tests can look for delays in your child's social and personal skills, fine and gross motor skills and language.

Parents are usually the first ones to think that there is a problem with their child's motor, social, and/or speech and language development, and this parental concern should be enough to initiate further evaluation. However even if you do not think you're your child is developmentally delayed regular assessment by a physiotherapist, occupational therapist and speech therapist are recommended for all children with VCFS due to the high incidence of developmental delay associated with this syndrome. Inclusion in early intervention programs have shown to have large benefits for VCFS child and reports from the above therapists will be necessary when applying for a position in such a program.

# **SPEECH AND LANGUAGE**

Speech-language impairment is one of the most common clinical features in velocardiofacial syndrome (VCFS). The most reported speech and language needs are: hypernasality, expressive language delays, articulation problems, auditory processing deficits, problem solving difficulties, reasoning difficulties, word finding problems, difficulty understanding idioms or words with multiple meanings and problems following multiple directions.

Hypernasality occurs when air escapes through the nose during the production of oral speech sounds resulting in reduced intelligibility. This is a common characteristic in the speech and language profile because 69% of children have palatal abnormalities. If the structure of the soft palate velum is such that it does not stop the flow of air from going up to the nasal cavity, it will cause hypernasal speech. This phenomenon is referred as velopharyngeal inadequacy (VPI). Hearing loss can also contribute to increased hypernasality because children with hearing impairments can have difficulty self monitoring their oral speech output.

Difficulties acquiring vocabulary and formulating spoken language (expressive language deficits) at the onset of language development is also common. Vocabulary acquisition is often severely delayed for preschool age children. In some recent studies, children had a severely limited vocabulary or were still nonverbal at 2-3 years of age. School age children do make progress with expressive language as they mature, but many continue to have delays and demonstrate difficulty when presented with language tasks such as verbally recalling narratives and producing longer and more complex sentences. Receptive language, which is the ability to comprehend, retain, or process spoken language, can also be impaired although not usually with the same severity as expressive language impairments.

Articulation errors are commonly present in children with VCFS. These errors include a limited phonemic (speech sound) inventory and the use of compensatory articulation strategies resulting in reduced intelligibility. Compensatory articulation errors made by this population of children include: glottal stops, nasal substitutions, pharyngeal fricatives, linguopalatal sibilants, reduced pressure on consonant sounds, or a combination of these symptoms. Of these errors, glottal stops have the highest frequency of occurrence. It is reasoned that a limited phonemic inventory and the use of compensatory articulation strategies is present due to the structural abnormalities of the palate. The speech impairments exhibited by this population are more severe during the younger ages and show a trend of gradual improvement as the child matures.

Your child should have regular follow-ups to assess and monitor their speech and language. A speech therapist is a wonderful source of fun and exciting ways to work speech therapy into your daily routine without your child noticing they are even doing it.

## **EDUCATION AND SCHOOLING SECTION**

Problems with education and schooling are probably the most common areas that parents worry about. It is essential that education be addressed from a very early stage, even before any problems are noted. Some VCFS children are able to go to main stream schools with learning support while others may need a more nurturing environment. Making this decision can be difficult. Ask your child's therapists and preschool teachers their opinion on your child's development and coping skills. This may help you decide where your child would be best placed.

Telling the school about your child's diagnosis is recommended. This way the school can monitor your child's progress and apply for funding grants to get extra services to help in areas your child has difficulties with.

Monitoring your child's progress from an early age and giving them intervention as soon as a problem starts to surface has been shown to give your child the best start possible.

## **EARLY INTERVENTION**

Early intervention is available for preschool aged children. You must apply in writing by June the year before your child is eligible. To apply you will need a current speech therapy assessment, developmental assessment and any other relevant reports such as OT or physiotherapy reports. Contact the Department of Education in your state to request an application form. They will inform you of your closest service and the date your application is due. Ring early to avoid missing the deadline. April or May gives you time to get the application and arrange any reports they require. Many research studies have been done on early intervention and all have shown the enormous benefits early intervention has on the children who have been part of the program.

## **PRESCHOOL**

This is a wonderful learning environment and it is highly recommended that VCFS children attend prior to school. Depending on your area and waiting lists you may need to put your child's name on a waiting list a few years before they are due to go. Let your preschool know that your child has VCFS and the specific issues that affect your child. The preschool can apply for special need funding. The reports are usually the same as what you need for the early intervention program.

Don't be afraid to ring a few preschools in your area and ask questions such as: Do they have a special needs teacher on staff? How do they use the funding you are eligible for? These are important years of development for your child and setting a good foundation for school is important. Ensuring that social skills are developing and social language is encouraged are important aspects during this time and will help give your child the skills they need when they start school.

## **SCHOOL**

Choosing the type of school that is best for your child can be a daunting task and you should start the search about two years before your child is due to start school. Ring your local schools and talk to them. Ask them what support they have for special needs children, what funding system they use, how big the school is, what are the class sizes, do they have reading recovery, are they open to your therapist coming into the classroom to assess your child's learning and offer support to the teacher. There are many things you can ask the school to help you decide the best place for your child. Different schools have different funding and services. In some schools P & F or P & C committees may fund extra support for special needs children. When you have selected your school invite them to be part of your child's IEP (individual education plan) meetings in the year leading up to them starting school. The school will also let you know what reports they need to apply for funding so that when your child starts school support is available from day one to help with a smooth transition.

Talk to the therapists, special needs teachers and preschool teachers regarding your child whether they believe your child will be capable of mainstream education or if a more nurturing environment may be more suitable. Many special needs school also run a

transition program into mainstream. Look at all your options and use the knowledge of support of those around you to help guide your decision.

## **INDIVIDUAL EDUCATION PLAN (IEP)**

The Individual Education Plan is a recognised means of developing and reviewing agreed educational goals for a student. All students (3-6 years) who are receiving support through early intervention programs and all school students identified as requiring learning support are required to have an IEP. The IEP process brings parents, professionals and the student (where appropriate) together as a team to consider the student's current level of performance and to determine needs and learning priorities for the next six months. Some team members may be heavily involved in the development of the IEP while others, e.g., audiologist or ophthalmologist, may provide specific information only.

The IEP process promotes:

- shared responsibility for decision making and programming
- consensus about educational goals for the student
- collective accountability for outcomes, and
- a communication channel.

The product of the IEP process is a plan which provides key information to assist educators in meeting a student's individual educational needs resulting from a disability. It is not a plan of total instruction for students with disabilities. Other curriculum areas are covered in the general programming of the class. The IEP forms part of the student's file which should accompany the student when transferring to another school.

## **SERVICES TO HELP WITH LEARNING NEEDS**

### **TUTORING**

Private tutoring is a great way of helping your child get the extra help they require with their schooling. Your local newspaper or shopping centre usually has advertisements for tutors. Some areas have learning centres which offer tutoring. Other parents in your area may also be able to give a verbal recommendation for a suitable person. Rates for this will vary.

### **LEARNING LINKS**

Learning Links is an Australian charity and non-profit organisation formed to help children who have learning disabilities, difficulties and developmental delays and their families. Their services include early childhood intervention, full cognitive, developmental and pediatric assessments, individualised and supplementary specialist literacy and numeracy services, specialised help for children who have had serious illnesses, an in-school reading program for children falling behind in literacy, occupational therapy, speech therapy, physiotherapy, family counseling, case management, group programs and professional development. The children they help range in age from birth to 18 years. For more information see the website below.

[www.learninglinks.org.au](http://www.learninglinks.org.au)

## **CHERI**

The Children's Hospital Education Research Institute (CHERI) is a unique research institute that aims to improve the interface between children's health and their education. CHERI is part of The Children's Hospital at Westmead.

CHERI conducts research into the educational and psychosocial aspects of children with learning problems specifically VCFS. They do this by running clinics which provide educational and psychosocial assessments of children, as well as family counselling. CHERI also provides information and resources to families and professionals through its conferences, research forums, and helpful information sheets.

VCFS information sheets are available for download on the CHERI website (<http://www.cheri.com.au/>). Topics include:

- Assistance for children with VCFS – starting out
- Fine motor coordination and writing skills in children with VCFS
- Helping children with VCFS cope better in the playground
- Playdates

Referrals to the CHERI clinic are generally made by staff of the Clinical Genetics Department of The Children's Hospital at Westmead. If your child has not attended this clinic, but would like to make an appointment for an assessment at CHERI, please contact CHERI who will assess your child's eligibility.

For more details on CHERI please visit their website at [www.cheri.com.au](http://www.cheri.com.au)

## **NSW DEPARTMENT OF EDUCATION AND TRAINING**

The website below is for the NSW department of education and training. It lists all your schooling options, explains funding and how it is used to help your child. It also gives contact details for you to contact the disability program coordinator.

<http://www.schools.nsw.edu.au/studentsupport/programs/disability.php>

## **LEAVING SCHOOL**

Some young adults with VCFS want to go on to further education. Tertiary institutions offer a wide variety of courses allowing them to choose an area of interest. TAFE offers many supported courses for individuals with varying disabilities. Before your child finishes school make a meeting with your school councillor or academic advisor to discuss suitable options that interest your child.

## **EMPLOYMENT**

Individuals with VCFS have different levels of functioning and different needs when obtaining employment. For some they may be able to maintain full-time employment while part-time or casual may be more appropriate for others. There are many agencies that help adults with disabilities obtain work. The following website gives a list of the community-based organisations that assist with the employment of people with a disability in NSW.

<http://www.eeo.nsw.gov.au/jobs/disabil/service.htm>

## **AS ADULTS**

We are still learning about how our children might be as adults. However, there are many parents out there who have the deletion and they were not aware of it until they had a child. Being very positive, this means that the outlook can be very good. Our children will hopefully develop strategies for dealing with problems they come across in life and we will be there to advise them when they need it.

## **MENTAL HEALTH ISSUES**

A number of young adults with VCFS have been found to suffer from mental health problems. The psychiatric studies that have been published until now have presented some conflicting information, but it is safe to say that the majority of individuals with VCFS have some behavioral disorders which often begin after puberty when hormonal changes occur in the body. In most of these cases, the manifestations are not very severe and often do not require medical management. The frequency of severe psychiatric problems such as schizophrenia and bipolar disorder is fairly low, probably under 20%, but this exact figure is not known. Part of the problem in psychiatric investigations is that the severity of psychiatric illness varies with age, typically becoming more apparent in adult life. However, enough adults with VCFS have not been studied to know the exact extent and severity of mental illness in this population.

# TELLING YOUR CHILD THEY HAVE VELO CARDIO FACIAL SYNDROME (22Q11 DELETION)

There is no fixed age and no fixed way to tell someone that they have VCFS. It depends very much on the individual's mental age, their level of understanding and their need to know. Telling an individual about the diagnosis is not completed in a single conversation. It is a long-term process and may bring out a variety of responses ranging from denial to relief. Specialised support and counselling may be required to assist the individual and others through this process.

There are a number of benefits in telling the individual and others about the diagnosis. These may include:

- Having a diagnosis and information about the VCFS can assist with understanding the feelings of being different
- Understanding the specific needs of people with VCFS provides an opportunity to focus on strengths related to the disability
- Explaining to teachers or fellow students can result in a better understanding of the individual's capabilities and facilitate greater inclusion. The same holds true for employment settings.

However, providing information about the diagnosis can result in an individual feeling vulnerable and being the subject of teasing, bullying and/or discrimination. Individual circumstances must be considered when it comes to telling others about the diagnosis.

## BENEFITS

You may be entitled to the following benefits from Centrelink.

Career allowance – a payment for the extra time it takes to look after an individual with a disability. There is no asset or income test.

Career payment – a payment for the extra time it takes to look after an individual with a disability. There is an asset and income test.

Disability support pension – for individuals over the age of 16 with a disability.

Health Care Card – usually comes with the above payments but can be applied for separately.

You may be entitled to the following benefits from Medicare.

Cleft Palate Dental Scheme – available to any individual with a cleft palate or lip. Gives the holder free dental care until the age of 28. Conditions apply to treatment.

The scheme provides Medicare benefits for dental treatment including:

- a limited range of orthodontic work
- surgical extraction of teeth by oral and Maxillofacial surgeons
- some general and prosthodontic services from your family dentist.

Allied Health Services/ Enhanced Primary care Plan – This benefit is part of the Chronic Disease Act. You need your GP to write an enhanced primary care plan to outline the specific needs that can be treated by an allied health professional. You can then have 5 free visits to allied health professional/s eg you may use 5 visits to the speech therapist or 2 to the speech therapist, 2 to an occupational therapist and 1 to a psychologist. The 5

visits can be used however you and your GP determine is best.

Psychological benefits – Your GP can refer to you for up to 12 free visits to a psychologist for treatment of mental health issues.

## **HOW DO I CLAIM THE ABOVE PAYMENTS?**

### Centrelink payments

You can download the claim forms online (<http://www.centrelink.gov.au/>) or ring 132717 for the forms to be posted to you. Complete the forms and return them to your nearest Centrelink Customer Service Centre.

### Medicare payments

You can download the claim forms online ([www.medicareaustralia.gov.au](http://www.medicareaustralia.gov.au)) or ring 132 011 or 1300 652 492 or visit your local Medicare office.

***“Genetics is not Destiny”***